Reviewer's report

Title: Rapid detection of PAH gene mutations in Chinese people

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Reviewer: Brigitte Glanzmann

Reviewer's report:

The authors describe a novel method for the accurate, reliable, fast and simple diagnosis of phenylketonuria (PKU). This autosomal recessive condition results in phenylalanine hydroxylase (PAH) deficiency which contributes to metabolic conditions. The authors describe a combination of single-tube PCR and molecular hybridization to identify PAH gene mutations. A total of 9 mutations were identified in Chinese individuals. The authors therefore conclude that this novel technique can be used in the Chinese population and suggest that it is suitable for the genetic diagnosis of other large populations affected by PKU.

This project has merit, but most of the paragraphs added into this manuscript have not been checked to ensure that the language conveys what the researchers are trying to get across to the reader. This needs to be addressed before this manuscript can be accepted.

One such example is illustrated below:

"Similarly it is suggested that the mutational site is the heterozygote if all wild-type control spots and some specific mutational site on the hybridized membrane strip present a clear blue spot. However, it is suggested that the test is fail while wild-type control spots and mutational spots are not colored simultaneously and it needs to be retested after the analysis of the cause".

* It is very difficult to ascertain what the authors are trying to get across to the readers.
Are the methods appropriate and well described?
If not, please specify what is required in your comments to the authors.

Yes

Does the work include the necessary controls?
If not, please specify which controls are required in your comments to the authors.

Yes

Are the conclusions drawn adequately supported by the data shown?
If not, please explain in your comments to the authors.

Yes

Are you able to assess any statistics in the manuscript or would you recommend an additional statistical review?
If an additional statistical review is recommended, please specify what aspects require further assessment in your comments to the editors.

Not relevant to this manuscript

Quality of written English
Please indicate the quality of language in the manuscript:

Needs some language corrections before being published

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